# Online testing tool identifies people likely to benefit from genetic testing for inherited risk for certain cancers

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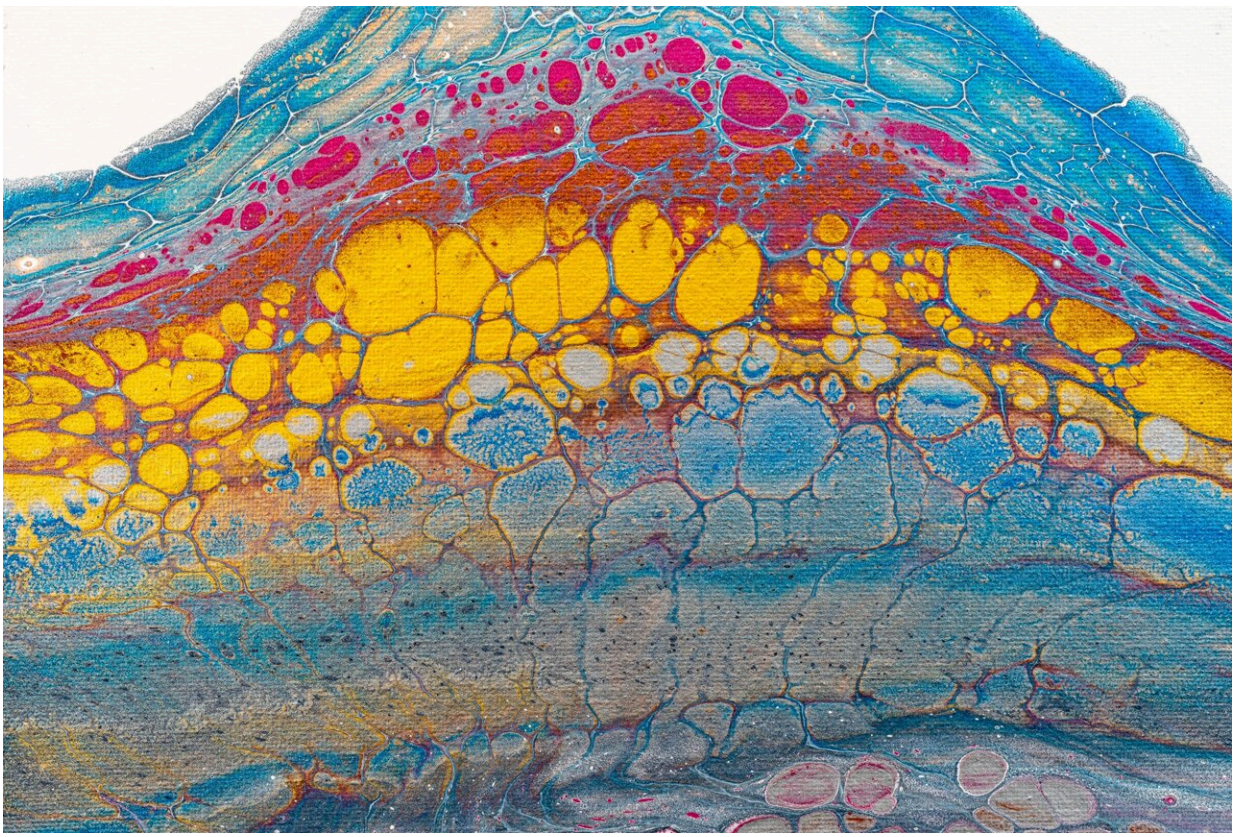

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An online tool developed by researchers and physicians at Dana-Farber Cancer Institute can accurately and rapidly identify people who should

undergo testing for inherited genetic changes that raise the risk of developing certain cancers, a new study shows.

The tool, known as PREMMplus, assesses whether an individual is likely to carry an inherited (or germline) change in nearly 20 genes linked to cancer. People with a high probability score could then receive genetic testing to determine if they harbor such changes and could benefit from measures to prevent the cancer or detect it at the earliest possible stage.

The tool fills a clear need at a time when the field of cancer genetic risk and genetic counseling is undergoing rapid change, say the authors of the study, published online today in the *Journal of Clinical Oncology*.

"Today, we know of many more inherited genetic variations associated with cancer than we knew of just 10 years ago, and it's now standard practice at many cancer centers to test patients and family members for these variations," says the senior author of the new study, Sapna Syngal, MD, MPH, of Dana-Farber and Brigham and Women's Hospital. "It has become increasingly complex to determine which individuals warrant germline testing. At the same time, genetic counselors—who traditionally meet with people prior to testing—are in short supply."

PREMMplus asks users questions about their age, sex, ethnicity, and personal and family history of 18 cancers. It then calculates the probability that the person carries an inherited mutation in any of 19 genes associated with those cancers. The researchers recommend that people whose probability is 2.5% or greater be referred for testing to verify if they have that mutation.

The PREMMplus model was developed using data from 7,280 adults who underwent testing for hereditary cancer risk at Dana-Farber's Genetics Program over a 15-year period. Genetic counselors interviewed each of the participants, collecting personal data and detailed family

histories of cancer occurrences. By correlating the genetic testing results with information gathered by counselors, Dana-Farber data scientists produced an algorithm for predicting the likelihood that an individual has an inherited mutation in any of the 19 cancer-related genes.

The current study tested the predictive power of PREMMplus in more than 30,000 people who had undergone germline testing for multiple cancer-susceptibility genes at Dana-Farber and California-based Ambry Genetics. Researchers found it had high sensitivity: the vast majority of users predicted to have a specific germline change actually did have that change, as shown by genetic testing. Likewise, it had a high negative-predictive value: when users were predicted not to have specific germline changes, genetic testing found that they did not, in fact, have them.

"Our findings show that PREMMplus has the potential to change the model by which patients and family members are referred for genetic testing and counseling," Syngal remarks. "Traditionally, if a physician or patient is concerned about a family history of cancer, the patient is referred to a genetics clinic, where a counselor takes a complete family history. PREMMplus can be directly embedded in a patient's electronic medical record, and patients could fill out the questionnaire themselves. Based on the result, health-care providers can then determine who should receive genetic testing.

"At a time when there's a shortage of genetic counselors, PREMMplus can help streamline risk assessment and ensure that their time can be focused on where they're most needed—helping people understand the results of genetic testing and the options available when a cancer -susceptibility gene is found."

**More information:** Matthew B. Yurgelun et al, Development and Validation of the PREMMplus Model for Multigene Hereditary Cancer

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